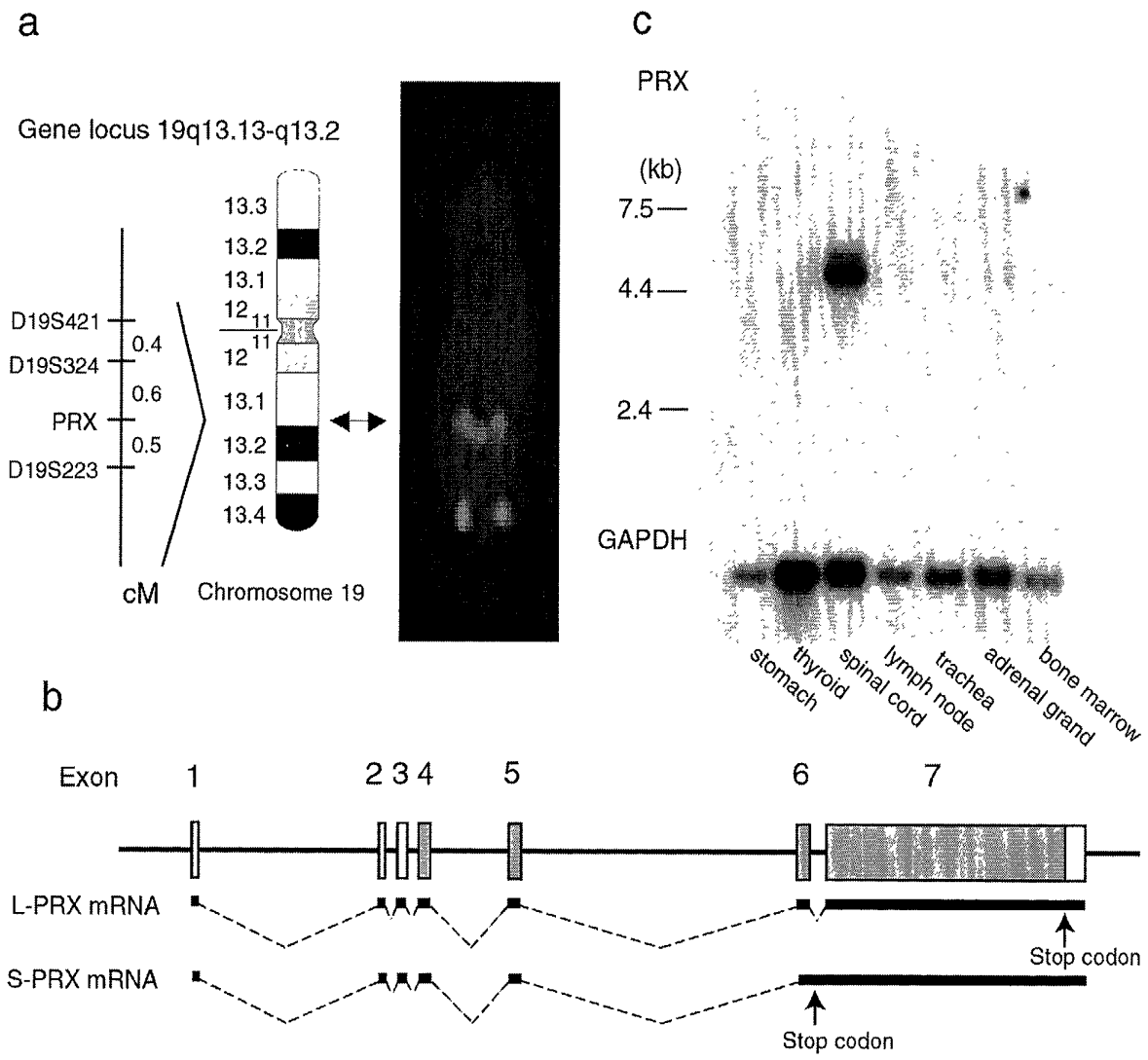


FIG. 1

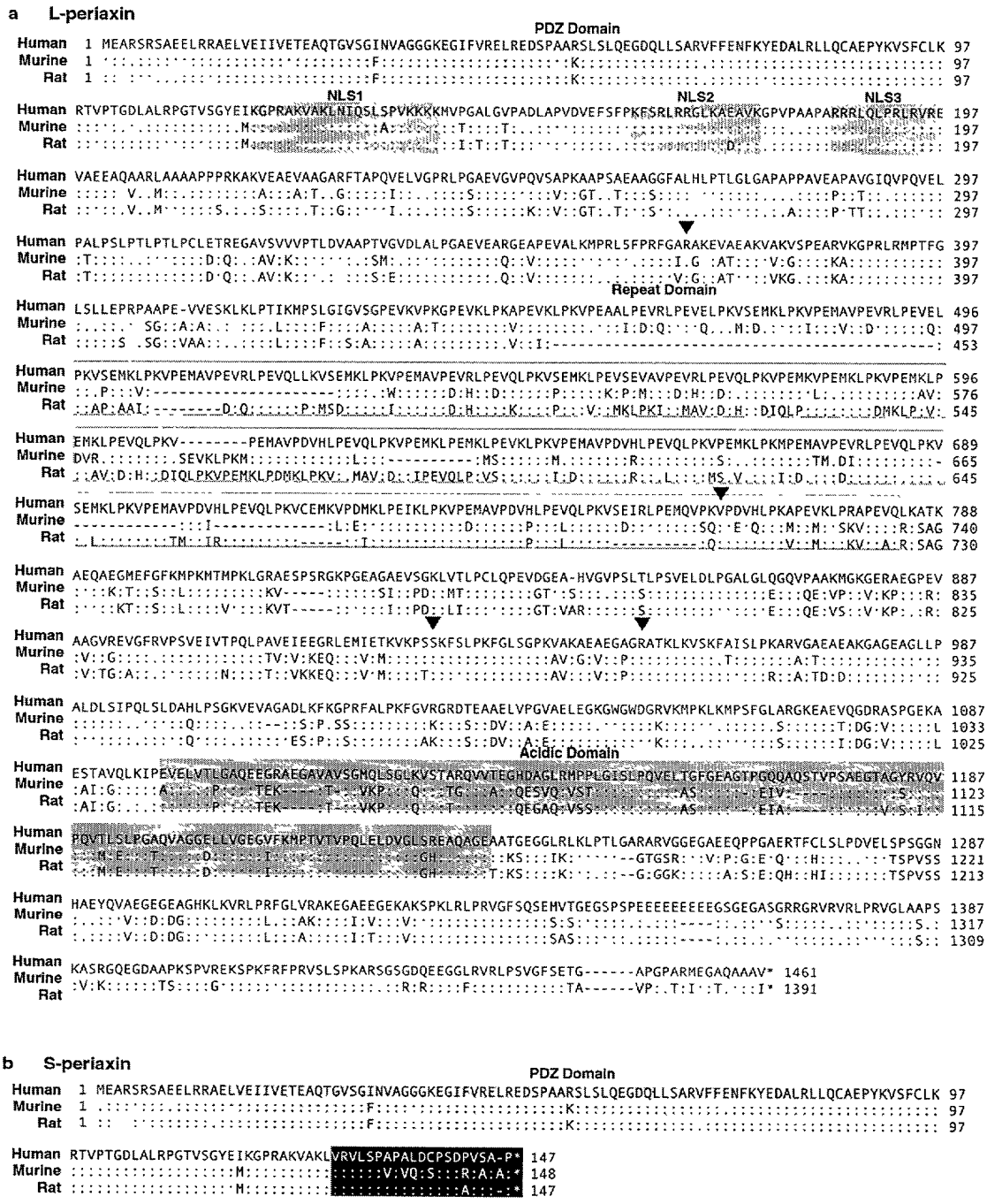

a L-periaxin
PDZ Domain
NLS1
NLS2
NLS3
Repeat Domain
Acidic Domain
b S-periaxin
PDZ Domain

FIG. 2

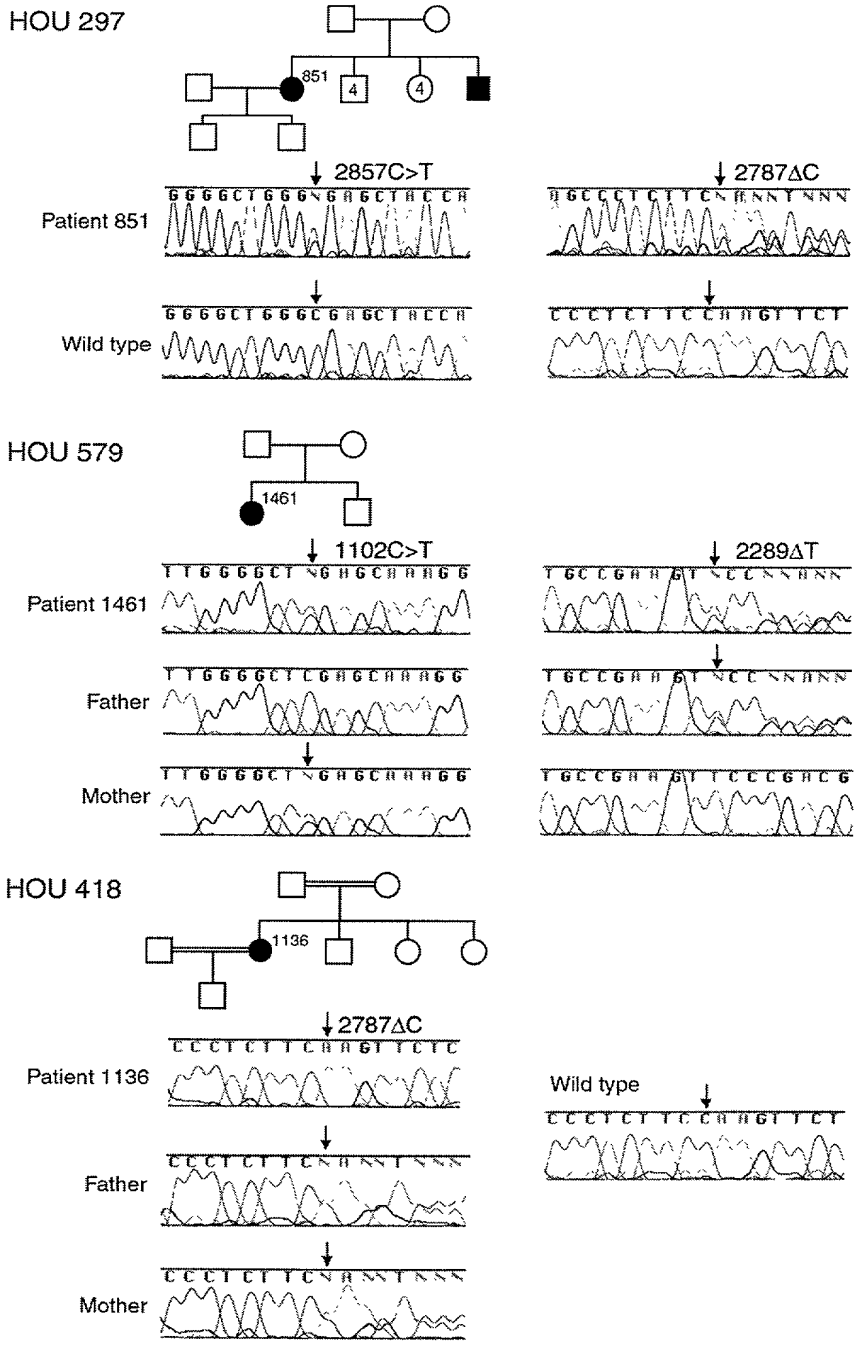
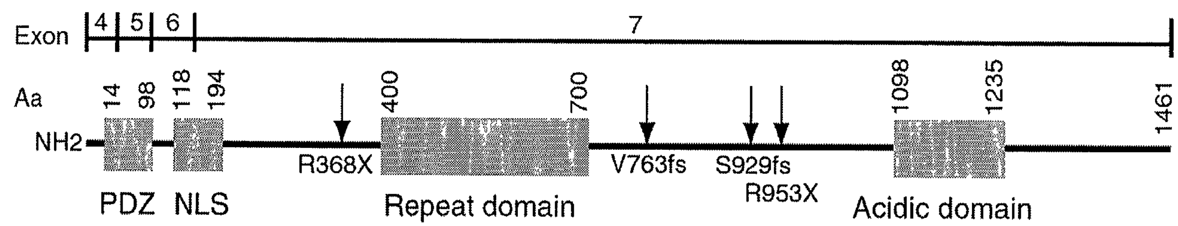

HOU 297
851
4
4
2857C>T
2787ΔC
Patient 851
Wild type
HOU 579
1461
1102C>T
2289ΔT
Patient 1461
Father
Mother
HOU 418
1136
2787ΔC
Patient 1136
Wild type
Father
Mother

FIG. 3



Patient	Phenotype	Pathogenic Allele 1	Frequency	Pathogenic Allele 2	Frequency
851	DSN	2857C>T R953X	0/190	2787ΔC S929fsX957	0/190
1461	DSN	1102C>T R368X	0/178	2289ΔT V763fsX774	0/176
1136	DSN	2787ΔC S929fsX957	0/190	2787ΔC S929fsX957	0/190

FIG. 4

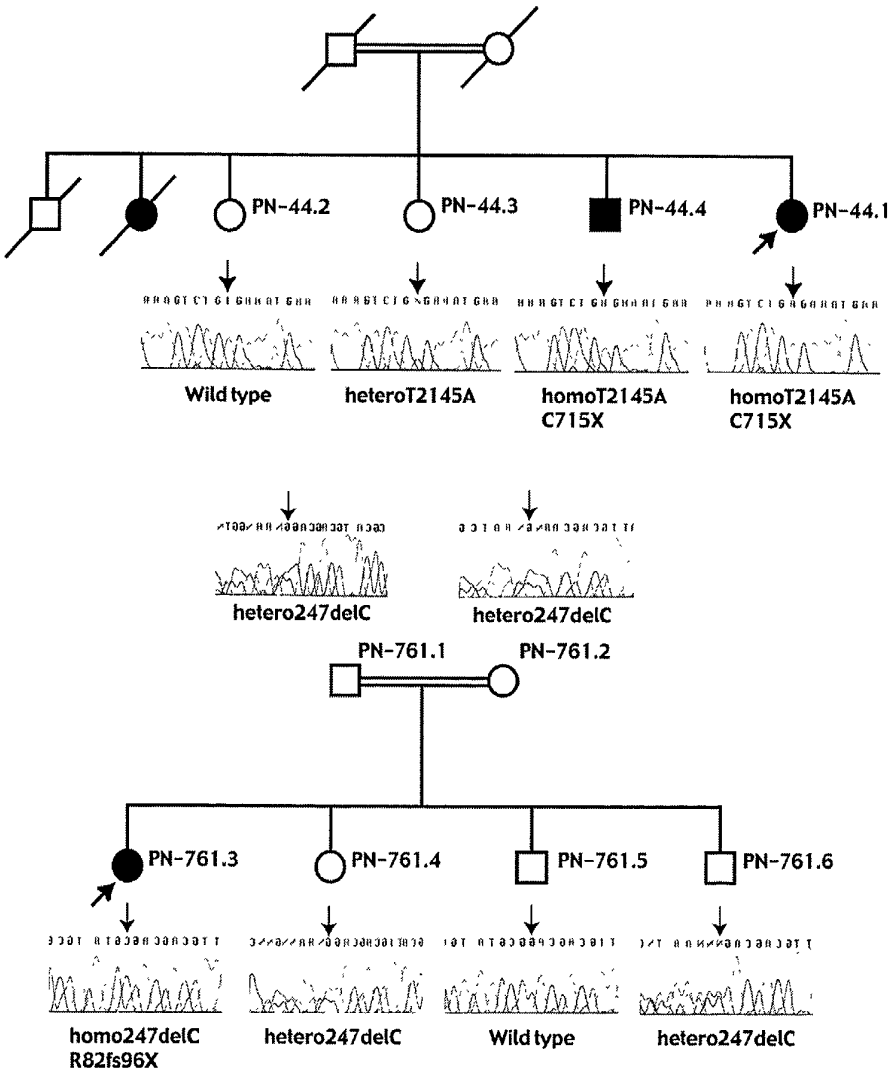

PN-44.2
PN-44.3
PN-44.4
PN-44.1
Wild type
heteroT2145A
homoT2145A
C715X
homoT2145A
C715X
hetero247delC
hetero247delC
PN-761.1
PN-761.2
PN-761.3
PN-761.4
PN-761.5
PN-761.6
homo247delC
R82fs96X
hetero247delC
Wild type
hetero247delC

FIG. 5

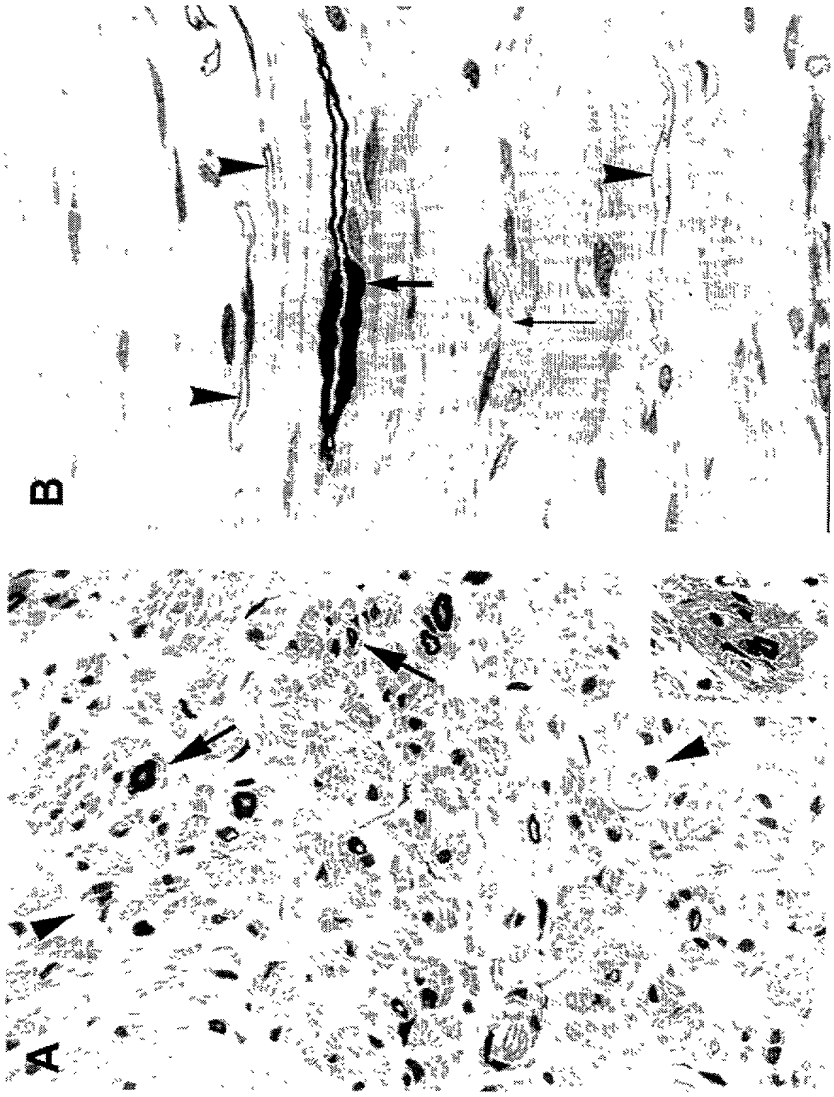


FIG. 6

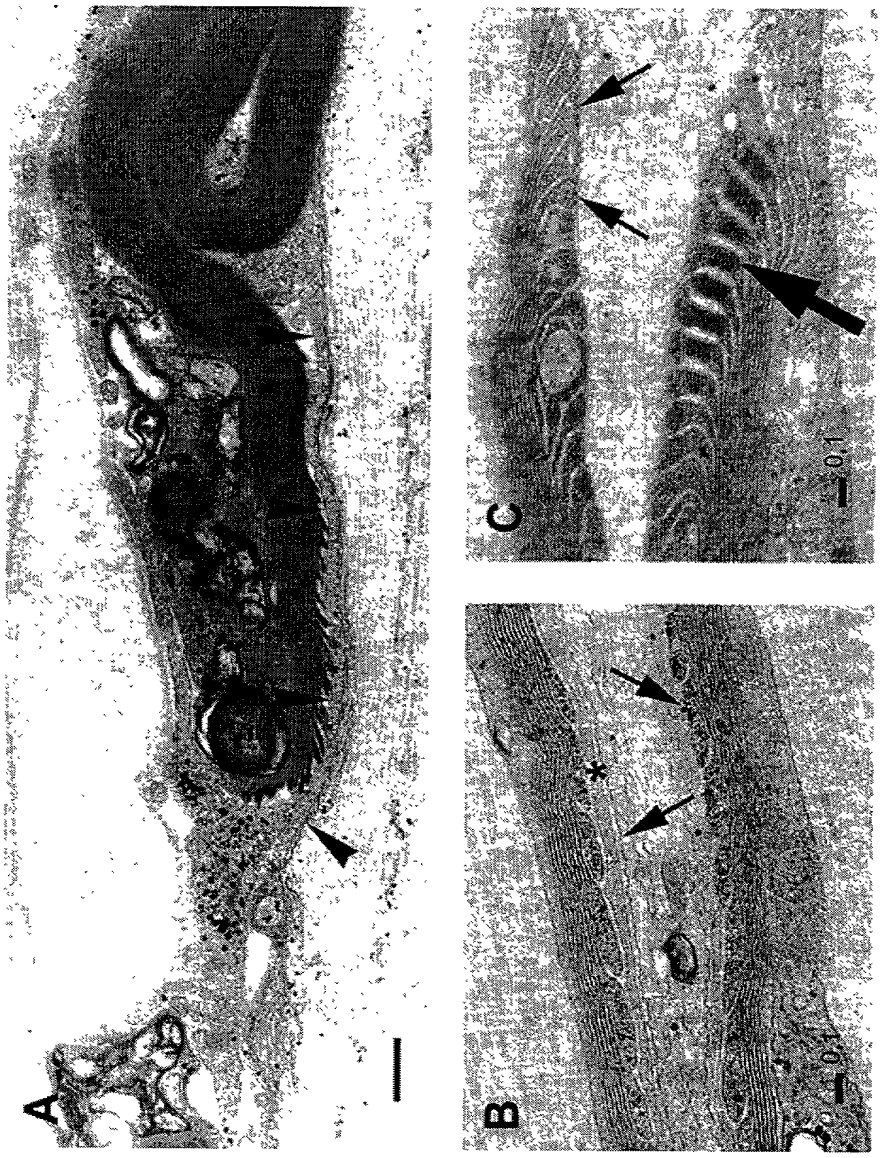


FIG. 7

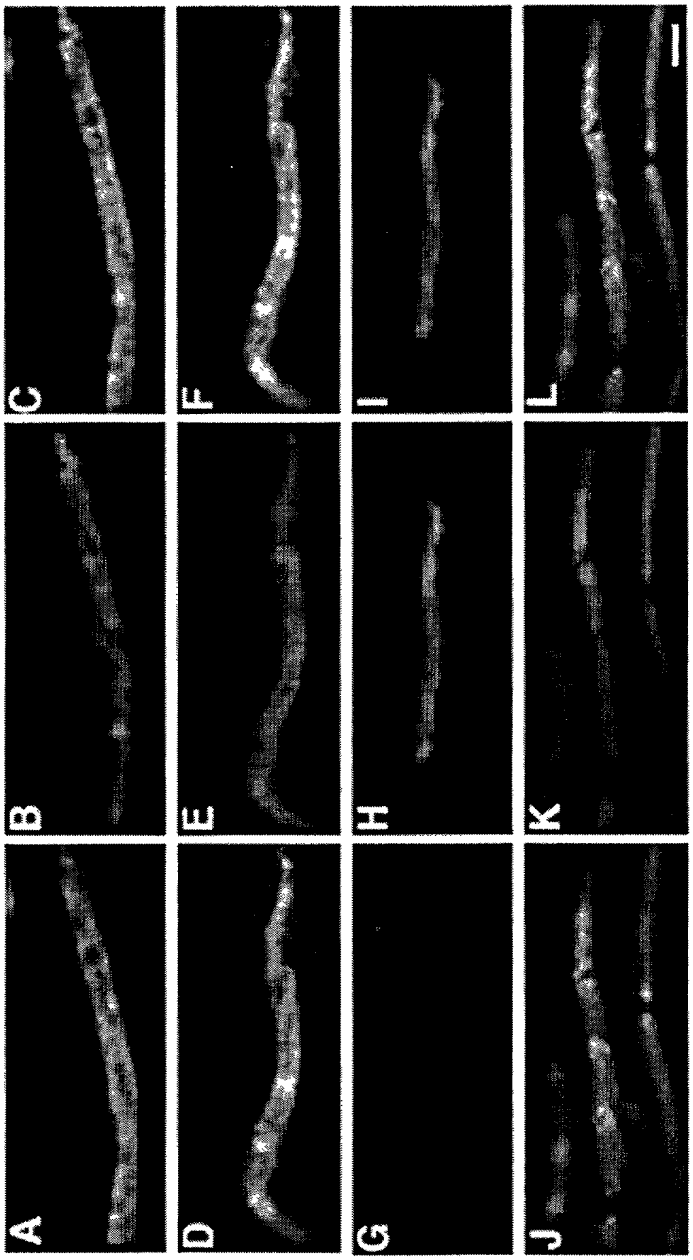
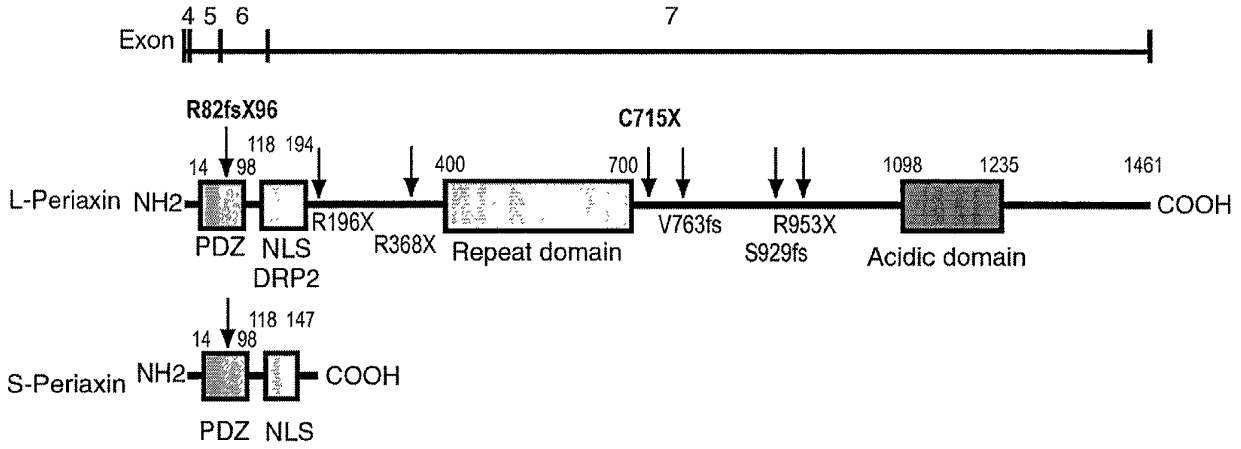


FIG. 8



Patient	Phenotype	Pathogenic Allele 1	Frequency	Pathogenic Allele 2	Frequency
PN44.1	CMT	C715X	0/180	C715X	0/180
PN44.4	CMT	C715X	0/180	C715X	0/180
PN761.3	CHN	R82fsX96	0/190	R82fsX96	0/190
851	DSN	R953X	0/190	S929fsX957	0/190
1461	DSN	R368X	0/178	V763fsX774	0/176
1136	DSN	S929fsX957	0/190	S929fsX957	0/190
CMT4F	DSN	R196X		R196X	

FIG. 9